

CHAPTER 4
BIRTH DEFECTS INSTITUTE
[Prior to 7/29/87, Health Department[470]]

641—4.1(136A) Program explanation. The birth defects institute within the department of public health consists of the Iowa neonatal metabolic screening program, the expanded maternal serum alpha-fetoprotein screening program, the regional genetic consultation service, the neuromuscular and related genetic disease program and the Iowa birth defects registry. The birth defects advisory committee represents the interests of the people of Iowa and assists in the development of programs that ensure the availability of and access to quality genetic health care services by all residents. The committee advises the director of the department of public health regarding issues related to genetics and hereditary and congenital disorders and makes recommendations about the design and implementation of the institute's programs. Committee membership is made up of representatives of professional groups, agencies, legislators, consumers and individuals with an interest in promoting genetic services for the residents of Iowa.

641—4.2(136A) Definitions. For the purposes of this chapter, the following definitions shall apply:

“*Central laboratory*” means the University Hygienic Laboratory.

“*Central registry*” means the Iowa birth defects registry.

“*Committee*” means the birth defects advisory committee.

“*Department*” means the Iowa department of public health.

“*Director*” means the director of the Iowa department of public health.

“*Institute*” means the birth defects institute within the Iowa department of public health.

641—4.3(136A) Iowa neonatal metabolic screening program. This program provides comprehensive newborn screening services for hereditary and congenital disorders for the state.

4.3(1) Newborn screening policy. All newborns shall be screened for hypothyroidism, phenylketonuria (PKU), galactosemia, hemoglobinopathies, congenital adrenal hyperplasia (CAH), and medium chain acyl Co-A dehydrogenase (MCAD) deficiency.

As new disorders are recognized and new technologies and tests become available, the institute shall follow protocols developed by the department in regard to the addition of disorders to or deletion of disorders from the screening panel. The state board of health shall provide final approval for the addition of new disorders to the screening panel.

4.3(2) Health care provider responsibility. The licensed attending health care provider shall ensure that infants under the provider's care are screened. A parent or guardian shall be informed of the type of specimen, how it is obtained, the nature of the disorders being screened, and the consequences of treatment and nontreatment. Should a parent or guardian refuse the screening, said refusal shall be documented in writing on the Iowa neonatal metabolic screening program waiver. The parent or guardian and licensed attending health care provider shall sign the waiver. The parent or guardian and the Iowa neonatal metabolic screening program shall be provided with a copy of the waiver. The original copy of the waiver shall become a part of the infant's medical record.

4.3(3) County registrar responsibility. When a parent or guardian visits a county registrar's office to register a birth that was not attended by a licensed health care provider, the county registrar shall inform the parent or guardian of the need for a blood test to screen for hereditary and congenital disorders.

4.3(4) Neonatal metabolic screening procedure.

a. Collection of specimens. A filter paper blood specimen shall be collected from the infant at least 24 hours after the infant's birth, but not later than five days after the infant's birth.

EXCEPTIONS:

(1) A blood specimen must be collected before any transfusion, even if the infant is less than 24 hours old.

(2) All infants shall be screened prior to discharge even if the infant is less than 24 hours old.

(3) An infant transferred to another medical facility must be screened by the receiving facility unless the infant has already been screened. The transferring facility is responsible for notifying the receiving facility of the status of metabolic screening.

(4) An exception to this time sequence shall be accepted for infants of parents or guardians informed by a county registrar of the need for metabolic screening.

b. Submission of specimens. All specimens shall be forwarded by first-class mail or other appropriate means within 24 hours after collection to the University Hygienic Laboratory, the institute's designated central laboratory.

c. Processing of specimens. The central laboratory shall process specimens within 24 hours of receipt. The central laboratory shall notify the submitting health care provider, birthing facility or drawing laboratory of an unacceptable specimen and the need for another specimen.

d. Reporting of presumptive positive test results. A presumptive positive test result shall be reported within 24 hours to the consulting physician, or the physician's designee, who shall then notify the attending health care provider. This initial report shall be followed by a written report to the attending health care provider and the birthing facility.

4.3(5) Consulting physician responsibility. Consulting physicians shall be designated by the institute in collaboration with the central laboratory to provide interpretation of test results and consultation to licensed health care providers. Under the direction of consulting physicians, follow-up programs shall be available for all individuals identified by newborn metabolic screening. The activities shall include consultation, treatment when indicated, case management, education and quality assurance. The follow-up programs shall submit an annual report to the institute summarizing these activities.

4.3(6) Central laboratory responsibility. The central laboratory shall:

a. Process specimens within 24 hours of receipt.

b. Notify the submitting health care provider, birthing facility or drawing laboratory of an unacceptable specimen and the need for another specimen.

c. Report a presumptive positive test result within 24 hours to the consulting physician or the physician's designee.

d. Distribute specimen collection forms, screening waivers, and other materials to birthing facilities.

e. Provide educational materials concerning specimen collection procedures.

f. Have available for review and provide to the department a written quality assurance policy covering all aspects of its newborn screening activity.

g. Submit semiannual and annual reports to the institute. These reports shall include (1) number of infants screened by birthing facility, (2) number of repeat screens by birthing facility, (3) number of presumptive positive results by disorder, (4) number of confirmed positive results by disorder, (5) number of rejected specimens by facility, (6) number of waivers received by facility, (7) results of quality assurance testing, and (8) screening activity, fiscal accounting and educational activity details.

h. Act as fiscal agent for program charges encompassing the analytical, technical, administrative, educational, and follow-up costs for the screening program.

4.3(7) Retention, use and disposition of neonatal metabolic screening specimens.

a. A neonatal metabolic screening specimen collection form consists of dried blood spots on filter paper and attached infant and birthing center information.

b. Specimen collection forms shall be held for one month in a locked area at the central laboratory. After one month, the forms shall be incinerated unless kept for program evaluation or research use.

c. Research use. Only anonymized specimens shall be made available for research purposes.

(1) An anonymized specimen is defined as one which cannot be traced back to or linked with the particular infant from whom the specimen was obtained. Specimens shall be anonymized by removing the dried blood spot portion from the infant information portion of the specimen collection form.

(2) Investigators shall submit proposals to use anonymized specimens to the committee. Any intent to utilize nonidentifiable information associated with the dried blood spot sample for the research study must be clearly delineated in the proposal.

(3) Before research can commence, proposals shall be approved by the appropriate human subjects review committees, the birth defects advisory committee, and the department.

4.3(8) Neonatal metabolic screening fee determination. Sixty days prior to the end of the fiscal year, the central laboratory and the consulting physicians shall submit a combined program proposal and budget to the institute for the coming year. The department shall annually review and determine the fee to be charged for all activities associated with this program. The review and fee determination shall be completed at least one month prior to the beginning of the fiscal year.

641—4.4(136A) Expanded maternal serum alpha-fetoprotein screening program. This program provides comprehensive expanded maternal alpha-fetoprotein screening services for the state.

4.4(1) Maternal screening policy. It shall be the policy of the state of Iowa that all pregnant women are offered expanded maternal serum alpha-fetoprotein screening. If a patient desires this screening test, the specimen shall be drawn and submitted by her health care provider to the University Hygienic Laboratory, the institute's designated central laboratory.

4.4(2) Expanded maternal serum alpha-fetoprotein screening procedure.

a. *Collection of specimens.* A serum or clotted blood specimen shall be collected from the patient during 15 to 20 weeks of gestation.

b. *Processing of specimens.* The central laboratory shall test specimens within three working days of receipt.

c. *Reporting of abnormal results.* Abnormal test results shall be reported within 24 hours to the consulting physician or the physician's designee who shall then notify the submitting health care provider. On the next working day, this initial report shall be followed by a written report to the submitting health care provider.

4.4(3) Consulting physician responsibility. A consulting physician shall be designated by the institute in collaboration with the central laboratory to provide interpretation of test results and consultation to the submitting health care provider. This physician shall provide consultation for abnormal test results, assist with questions about management of identified cases, provide education and assist with quality assurance measures. The screening program with assistance from the consulting physician shall submit semiannual and annual reports to the institute detailing program activities.

4.4(4) Central laboratory responsibility. The central laboratory shall:

a. Test specimens within three working days of receipt.

b. Distribute specimen collection kits and other materials to health care provider offices and drawing facilities as required.

c. Inform the submitting health care provider or drawing facility of an unacceptable specimen and request another specimen.

d. Provide educational materials concerning specimen collection procedures.

e. Have available for review a written quality assurance program covering all aspects of its screening activity.

f. Submit a monthly report detailing screening activity to the consulting physician. This report shall include (1) number of initial tests, (2) number of repeat tests, and (3) results of quality assurance testing.

g. Act as a fiscal agent for program charges encompassing the analytical, technical, administrative, educational and follow-up costs for the screening program.

4.4(5) Expanded maternal serum alpha-fetoprotein screening fee determination. Sixty days prior to the end of the fiscal year, the central laboratory and the consulting physician shall submit a combined program proposal and budget to the institute for the coming year. The department shall annually review and determine the fee to be charged for activities associated with this program. The review and fee determination shall be completed at least one month prior to the beginning of the fiscal year.

641—4.5(136A) Regional genetic consultation service (RGCS). This program provides comprehensive genetic services statewide through outreach clinics.

4.5(1) Provision of comprehensive genetic services. The department shall contract with the Division of Medical Genetics within the Department of Pediatrics at the University of Iowa to provide genetic health care and education outreach services for individuals and families within Iowa. The contractor shall provide semiannual and annual reports to the department as specified in the contract.

4.5(2) Clinic services. The services provided may include, but are not limited to: diagnostic evaluations, confirmatory testing, consultation by board-certified geneticists, genetic counseling, medical and case management, and referral to appropriate agencies.

4.5(3) Patient fees. A sliding fee scale for clinical services shall be established for patients attending the outreach clinics. The parameters for the sliding fee scale shall be based on federally established percent of poverty guidelines and updated annually.

Families/clients seen in the regional genetic consultation service clinics shall have bills submitted to third-party payers where applicable. Families/clients shall be billed on a sliding fee scale after third-party payment is received. Payments received from receipts of service based on the sliding fee scale or from the third-party payers shall be used only to support the RGCS.

641—4.6(136A) Neuromuscular and other related genetic disease program (NMP). This program provides comprehensive services statewide for individuals and families with neuromuscular disorders through outreach clinics.

4.6(1) Provision of comprehensive services. The department shall contract with the Department of Pediatrics at the University of Iowa to provide neuromuscular health care, case management and education outreach services for individuals and families within Iowa. The contractor shall provide semiannual and annual reports to the department as specified in the contract.

4.6(2) Clinical services. The services provided may include, but are not limited to: diagnostic evaluations, confirmatory testing, physical therapy, consultation by board-certified neurologists, genetic counseling, medical and case management, supportive services and referral to appropriate agencies.

4.6(3) Patient fees. A sliding fee scale for clinical services shall be established for patients attending the outreach clinics. The parameters for the sliding fee scale shall be based on federally established percent of poverty guidelines and updated annually.

Families/clients seen in neuromuscular outreach clinics shall have bills submitted to third-party payers where applicable. Families/clients shall be billed on a sliding fee scale after third-party payment is received. Payments received from receipts of service based on the sliding fee scale or from the third-party payers shall be used only to support the neuromuscular outreach clinics.

641—4.7(136A) Iowa birth defects registry. The Iowa birth defects registry provides active birth defect surveillance statewide.

4.7(1) Definition. Birth defects shall be defined as any structural or genetic abnormality that may adversely affect a child's health and development. The abnormality must be diagnosed or its signs and symptoms must be recognized within the first year of life.

4.7(2) Birth defects surveillance policy. Birth defects occurring in Iowa are reportable conditions and records of these birth defects shall be abstracted pursuant to 641—1.3(139A) and maintained in a central registry.

Birth defects surveillance shall be performed in order to determine the occurrence and trends of birth defects, to conduct thorough and complete epidemiological surveys, to assist in the planning for and provision of services to children with birth defects and their families, and to identify environmental and genetic risk factors for birth defects.

4.7(3) Central registry activities.

a. The institute shall establish an agreement with the University of Iowa to implement the activities of the central registry.

b. The central registry shall use the birth defects coding scheme defined by the Centers for Disease Control and Prevention (CDC) of the United States Public Health Service.

c. The central registry staff shall review hospital records, clinical charts, physician's records, vital records and prenatal records pursuant to 641—1.3(139A) and any other information that the central registry deems necessary and appropriate for birth defects surveillance.

d. A reportable defect occurring in a fetal death or pregnancy termination may be included in the central registry.

4.7(4) Department responsibility.

a. When a live infant's medical records are ascertained by the central registry, the department or its designee shall inform the parent or legal guardian by letter that this information has been collected and provide the parent or guardian with information about services for which the child and family may be eligible.

b. The institute and the central registry shall annually release aggregate medical and epidemiological information to medical personnel and appropriate state and local agencies for the planning and monitoring of services for children with birth defects.

4.7(5) Confidentiality and disclosure of information. Reports, records, and other information collected by or provided to the Iowa birth defects registry relating to a person known to have or suspected of having a birth defect are confidential records pursuant to Iowa Code section 22.7.

Personnel of the central registry and the department shall maintain the confidentiality of all information and records used in the review and analysis of birth defects, including information which is confidential under Iowa Code chapter 22 or any other provisions of state law.

Central registry personnel are authorized pursuant to 641—1.3(139A) to gather all information relevant to the review and analysis of birth defects. This information may include, but is not limited to, hospital records, physician's records, clinical charts, birth records, death records, fetal death records, prenatal records, vital records, and other reports relevant and necessary for birth defects surveillance.

No individual or organization providing information to the Iowa birth defects registry in accordance with this rule shall be deemed or held liable for divulging confidential information.

4.7(6) Access to information in the central registry. The central registry and the department shall not release confidential information except to the following, under the following conditions:

a. The parent or guardian of an infant or child for whom the report is made and who can demonstrate that the parent or guardian has received the notification letter.

b. A local birth-to-three coordinator or an agency under contract with the department to administer the children with special health care needs program, upon receipt of written consent from the parent or guardian of the infant or child.

c. A local health care provider, upon receipt of written consent from the parent or guardian of the infant or child.

d. A representative of a federal or state agency, to the extent that the information is necessary to perform a legally authorized function of that agency. The information provided may not include the personal identifiers of an infant or child with a reportable birth defect.

e. Research purposes.

(1) All proposals for research using the central registry data to be conducted by persons other than program staff shall first be submitted to and accepted by the researcher's institutional review board. Proposals shall then be reviewed and approved by the department and the central registry's executive committee before research can commence.

(2) The central registry shall submit to the Iowa birth defects registry's executive committee for approval a protocol describing any research conducted by the registry in which the registry deems it necessary to contact case subjects and controls.

These rules are intended to implement Iowa Code chapter 136A.

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